# Your Guide to Understanding Genetic Conditions

# DRD5 gene

dopamine receptor D5

#### **Normal Function**

The *DRD5* gene provides instructions for making a protein called dopamine receptor D5, which is found in the brain. This protein works together with a chemical messenger (neurotransmitter) called dopamine. Dopamine fits into the D5 receptor like a key in a lock, which triggers chemical reactions within nerve cells. Dopamine signaling has many critical functions in the brain, including regulation of attention, mood, memory, learning, and movement.

# **Health Conditions Related to Genetic Changes**

# benign essential blepharospasm

Several studies have examined a possible relationship between a common variation (polymorphism) in the *DRD5* gene and benign essential blepharospasm. The results of these studies have been mixed. Some research has suggested that the polymorphism, a short repeated segment of DNA known as allele 2, occurs more often in people with benign essential blepharospasm than in people without the disorder. However, other studies have found no connection between this polymorphism and benign essential blepharospasm. Researchers are still working to clarify whether variants in the *DRD5* gene are associated with this disorder.

## other disorders

Other polymorphisms in the *DRD5* gene appear to be associated with a common behavioral condition called attention deficit hyperactivity disorder (ADHD). This condition, which typically begins in childhood, is characterized by overactivity, impulsive behavior, and difficulty paying attention.

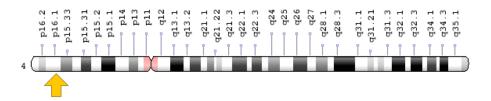
Most studies of the *DRD5* gene and ADHD have focused on a polymorphism located near the beginning of the gene. The region consists of two DNA building blocks (base pairs) that are repeated multiple times in a row. The size of this segment ranges from 134 to 156 base pairs. Multiple studies have suggested that a particular variant, which is 148-base pairs long, is associated with a moderately increased risk of ADHD. However, it is unclear how this polymorphism affects the risk of the disorder.

Variations in the *DRD5* gene are among many factors under study to help explain the causes of ADHD. A large number of genetic and environmental factors, most of which remain unknown, likely determine the risk of developing this complex condition.

## **Chromosomal Location**

Cytogenetic Location: 4p16.1, which is the short (p) arm of chromosome 4 at position 16.1

Molecular Location: base pairs 9,781,634 to 9,784,009 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

#### Other Names for This Gene

- d(1B) dopamine receptor
- d(5) dopamine receptor
- D1beta dopamine receptor
- DBDR
- dopamine D5 receptor
- dopamine receptor D1B
- DRD1B
- DRD1L2
- DRD5 HUMAN
- MGC10601

#### **Additional Information & Resources**

### **Educational Resources**

 Basic Neurochemistry (sixth edition, 1999): Dopamine Receptors https://www.ncbi.nlm.nih.gov/books/NBK27980/

#### Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DRD5%5BTIAB%5D%29+OR+%28dopamine+receptor+D5%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

#### OMIM

- ATTENTION DEFICIT-HYPERACTIVITY DISORDER http://omim.org/entry/143465
- DOPAMINE RECEPTOR D5 http://omim.org/entry/126453

#### Research Resources

- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=DRD5%5Bgene%5D
- HGNC Gene Family: Dopamine receptors http://www.genenames.org/cgi-bin/genefamilies/set/181
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene\_symbol\_report?q=data/ hgnc\_data.php&hgnc\_id=3026
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/1816
- UniProt http://www.uniprot.org/uniprot/P21918

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